



Considering rare diseases data in the EU health data strategy

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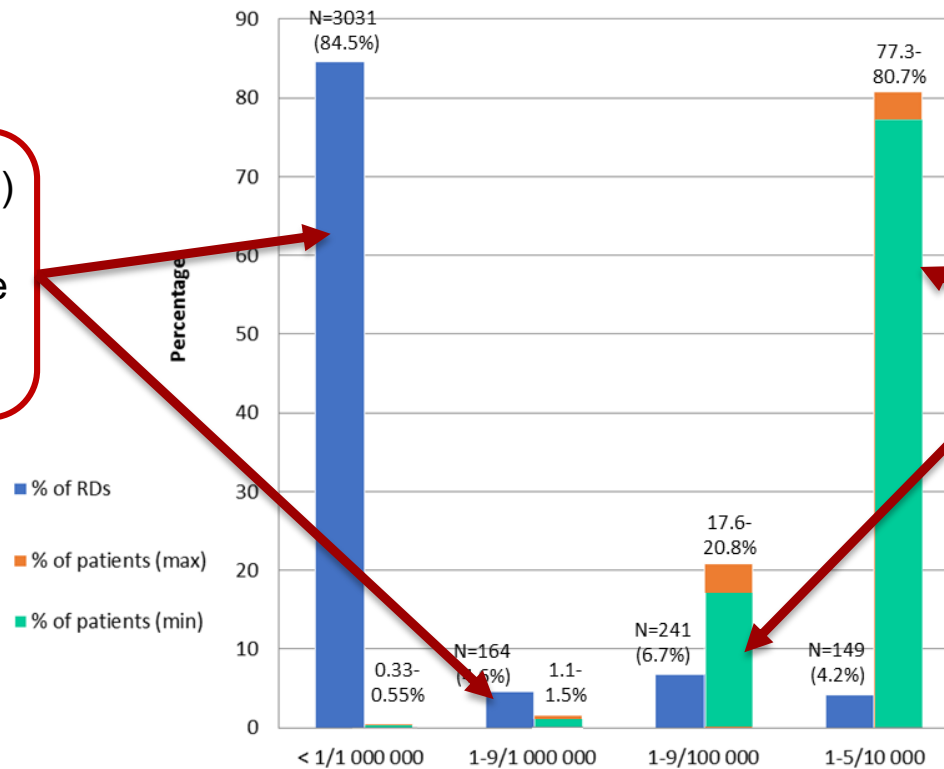
orphanet

 **Inserm**
La science pour la santé
From science to health

30 M+ European people matter

Where are they?

Most (89.1%) of rare diseases are very rare



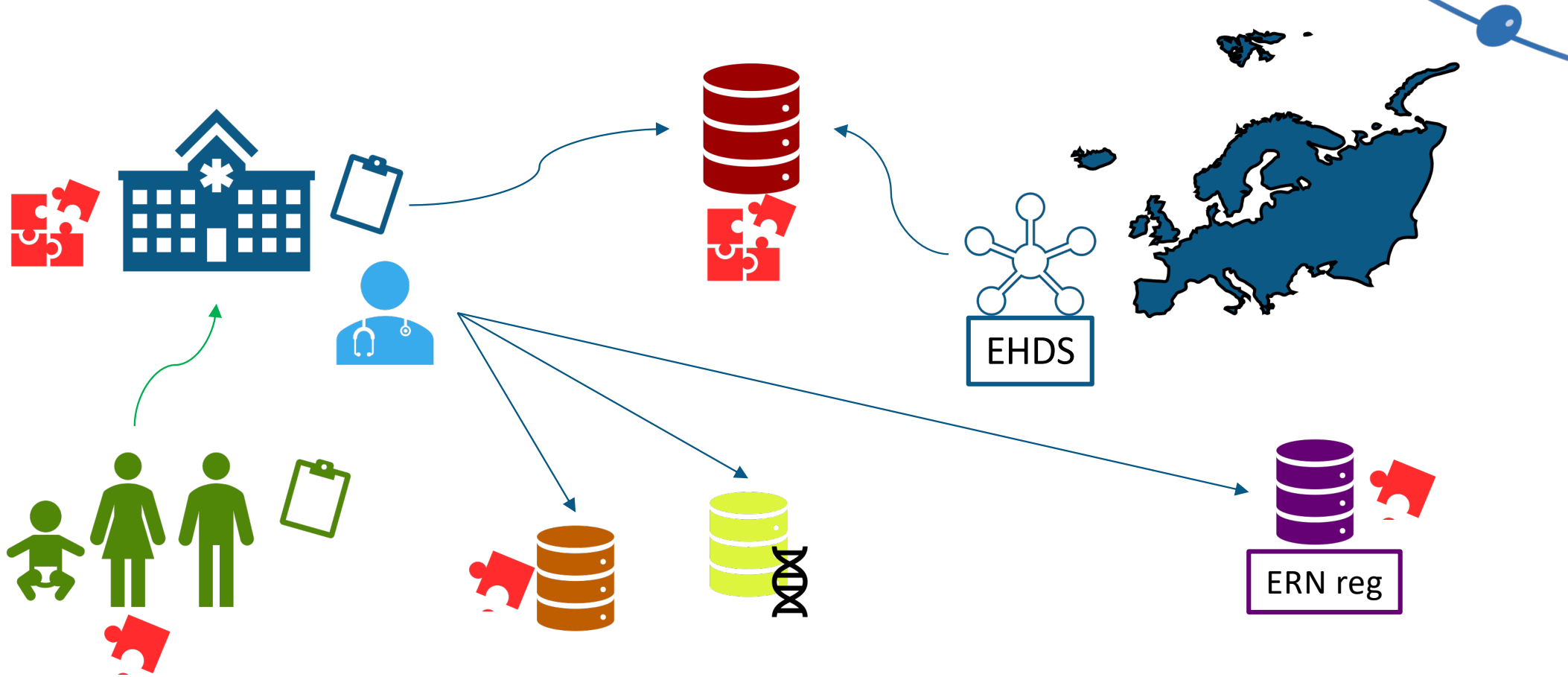
Most of the people with rare disease (>98%) have one of the 390 most prevalent diseases

3.5 - 5.9% of the population (263 - 446 Million people) worldwide

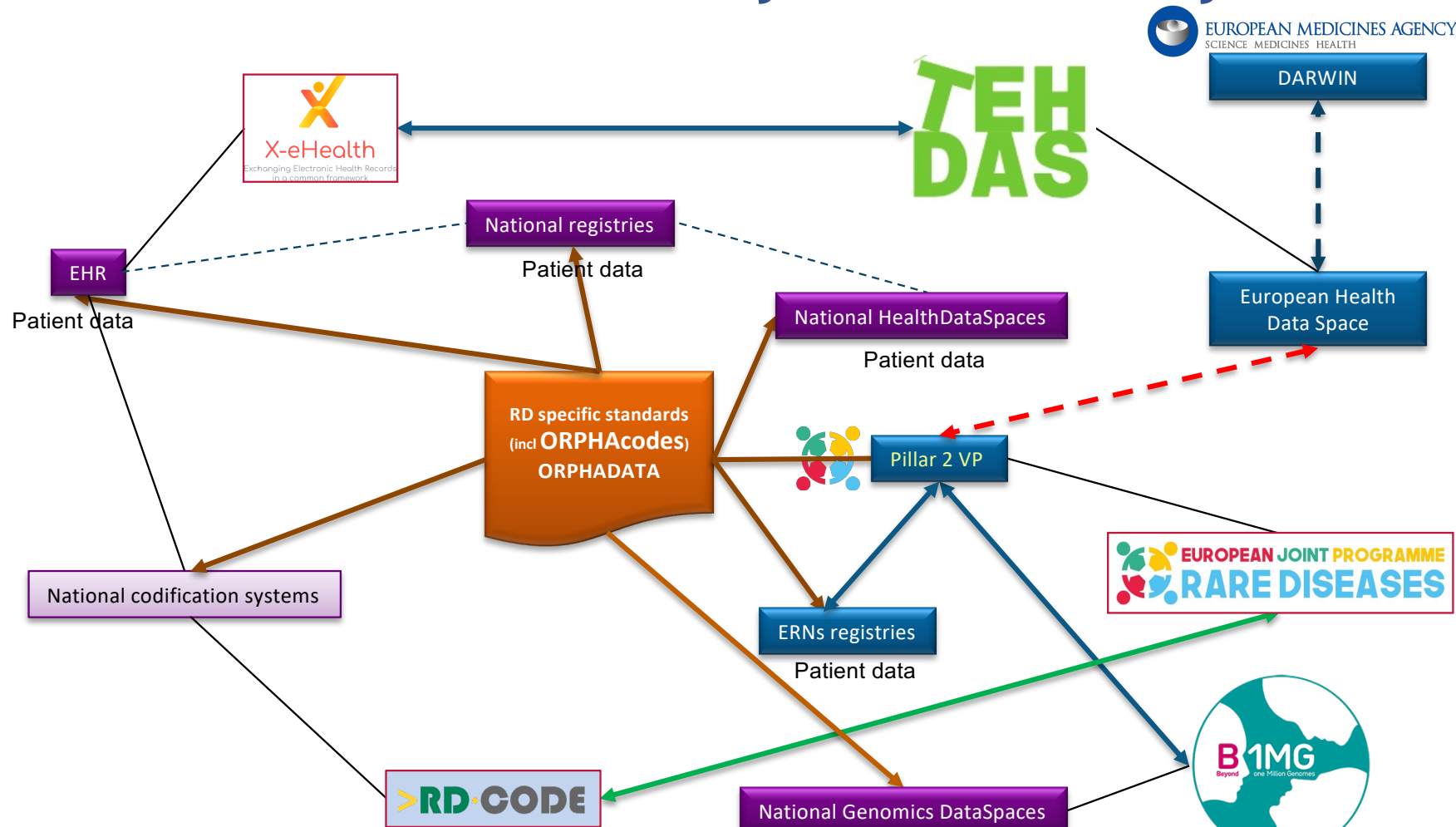
Eur J Hum Genet **28**, 165–173 (2020). <https://doi.org/10.1038/s41431-019-0508-0>

*Based on 68% of prevalent RD based on EU definition (<50/100,000), data from literature.

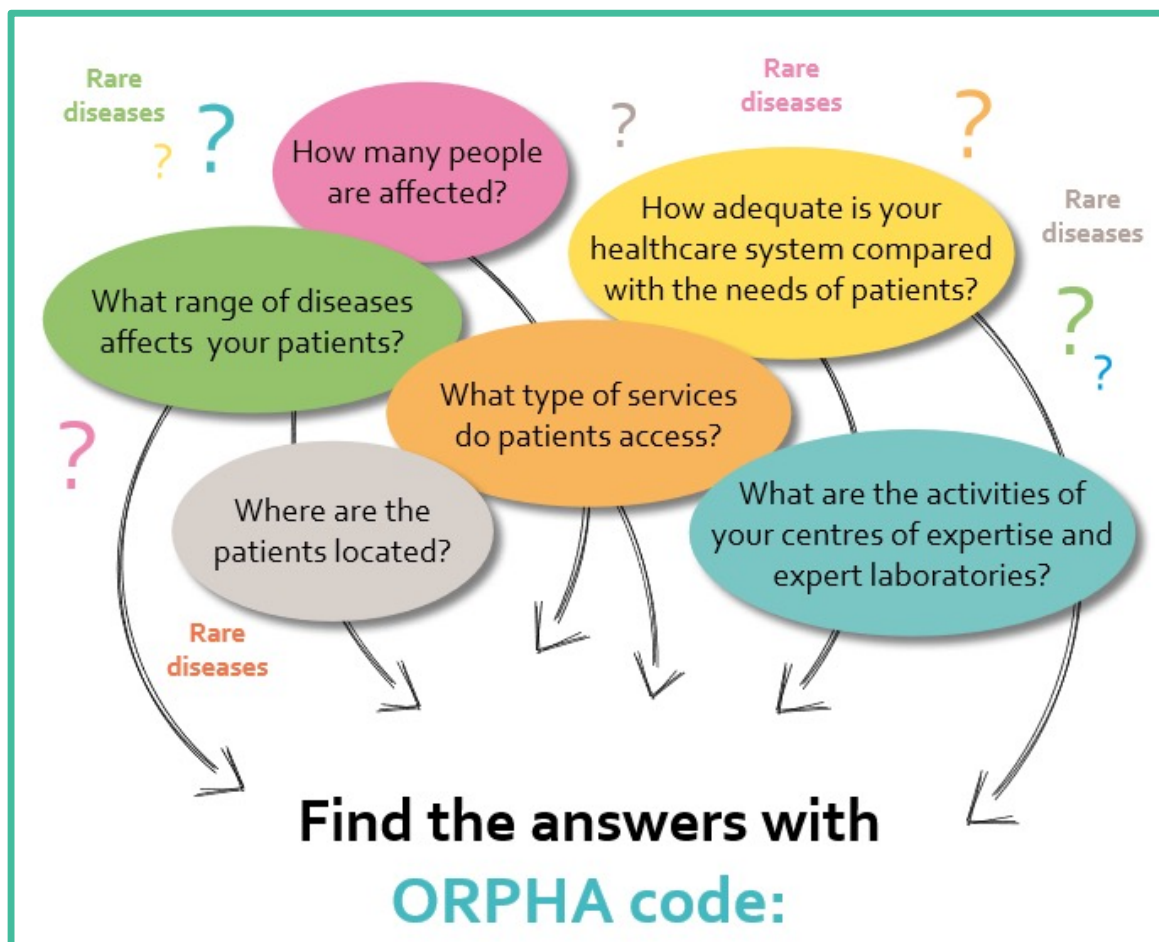
The (RD) data journey



Towards a seamlessly RD data ecosystem?



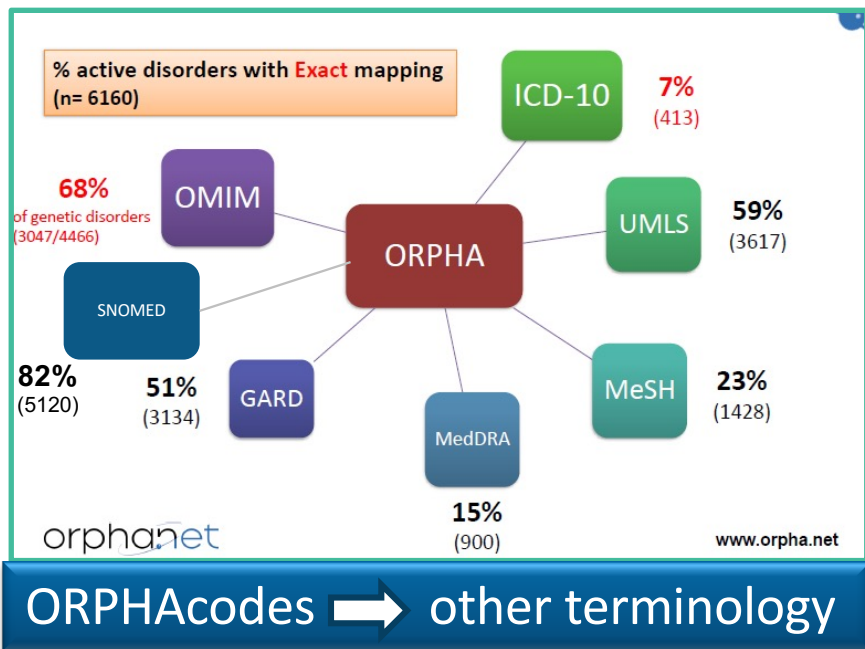
WHY ORPHAcoding



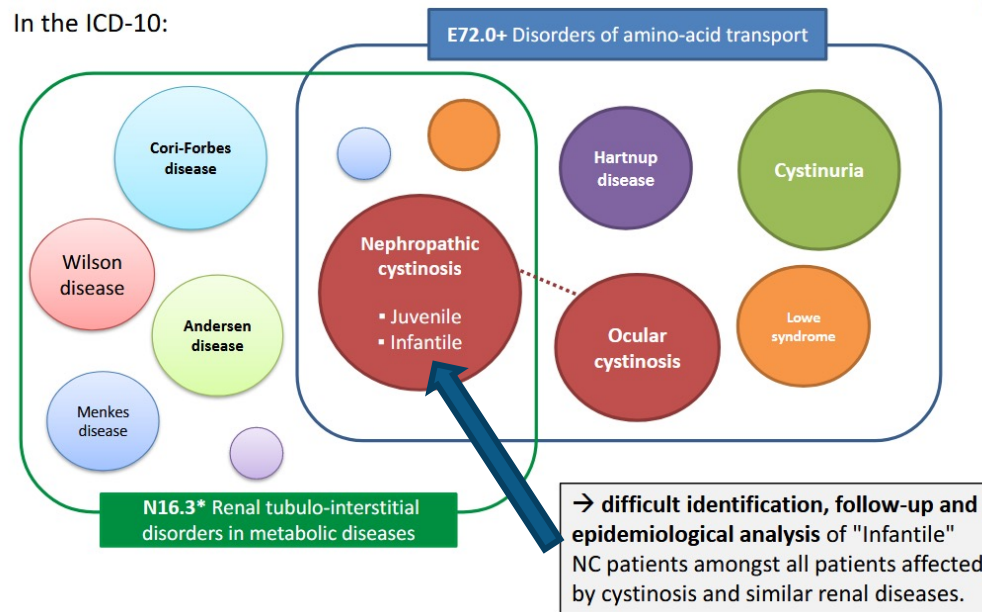
<p>Better care</p> <ul style="list-style-type: none"> • Improving knowledge on RD • Better evaluation of patients • Better referrals • Good practices • Emergency situations • Anaesthesia • Anticipate compensation for disabilities
<p>Evidence generation</p> <ul style="list-style-type: none"> • To answer a range of public health and research questions and make evidence based decisions

WHY ORPHAcoding

[ONLY ORPHACodes COVER PRECISELY ALL KNOWN RDs]



In the ICD-10:



6,000+ RDs Aligned with other terminologies to aid data interoperability

Ex. in Western Australia: the tip of the iceberg

The authors developed a resource set to interrogate the Western Australian Hospital Morbidity Data Collection between 1999 and 2010 and investigate :

THE TYPE OF RARE DISEASES REPORTED

UTILIZATION OF PARTICULAR SERVICES

THE NUMBER OF PATIENTS WITH RD IN WESTERN AUSTRALIA

THE COST OF RD TO THE WESTERN AUSTRALIAN HEALTH SYSTEM

DATASET ARE RICH ... BUT NOT DIRECTLY RELIABLE IN TERMS OF RD CLASSIFICATION



ORPHA CODES PROVIDES A BETTER RECOGNITION OF RD ... BUT IS NOT USED IN THE WESTERN AUSTRALIAN DATASETS

The authors matched Orpha Codes with the international classification of diseases system (ICD-10-AM) used in the database to create the **RD resource set** used to interrogate the database.

Data linkage with other administrative databases was made in order to avoid mismatches or duplicates.

MAIN FINDINGS

1

OVER REPRESENTATION OF PEOPLE WITH RD IN HOSPITAL DISCHARGES

In 2010, cohort members represented approximately 2% of the population of WA but accounted for 4.6% of people discharged and 9.9% of hospital discharges.

2

COHORT MEMBERS HAD A GREATER LENGTH OF STAY

People with RD had an average length of stay of 3.6 days in 2010 and 5.5 days when related to RD against 2.9 in the average population.

3

RD ARE ASSOCIATED WITH HIGHER COST FOR THE HEALTH SYSTEM

The total cost of hospital discharges for the cohort represented 10.5% of 2010 state inpatient hospital costs (4.6% for RD-related hospital discharges).

Walker, C., Mahede, T., Davis, G. *et al.* The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort. *Genet Med* **19**, 546–552 (2017). <https://doi.org/10.1038/gim.2016.143>

Why ORPHAcoding

EXAMPLE OF FRENCH NATIONAL RD REGISTRY > INDICATORS FOR NATIONAL PLAN

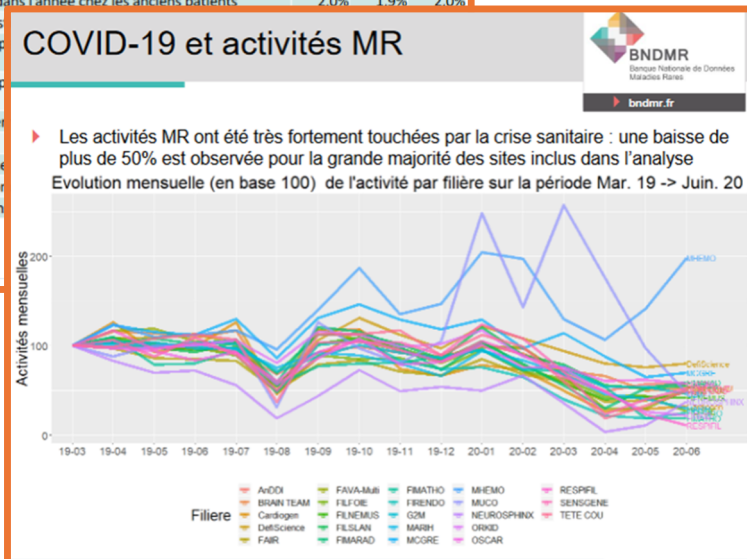


In France

- ORPHACodes are used in RD expert centres (EHR's specific RD section including the RD MDS)
- Deidentified data is nationally collected in the BNDMR the National RD registry
- https://www.bndmr.fr/wp-content/uploads/2020/02/MDS_v1.11-2EN.pdf

Indicateurs PNMR3

Indicateur	2018	2019	2020 (avril)
Proportion de patients de la file active adressés par des médecins libéraux aux CRMR et CCMR	13,4%	14,6%	14,4%
Proportion de diagnostics réalisés dans l'année chez les nouveaux patients	17,8%	17,9%	17,9%
Proportion de diagnostics réalisés dans l'année chez les anciens patients	2,0%	1,9%	2,0%



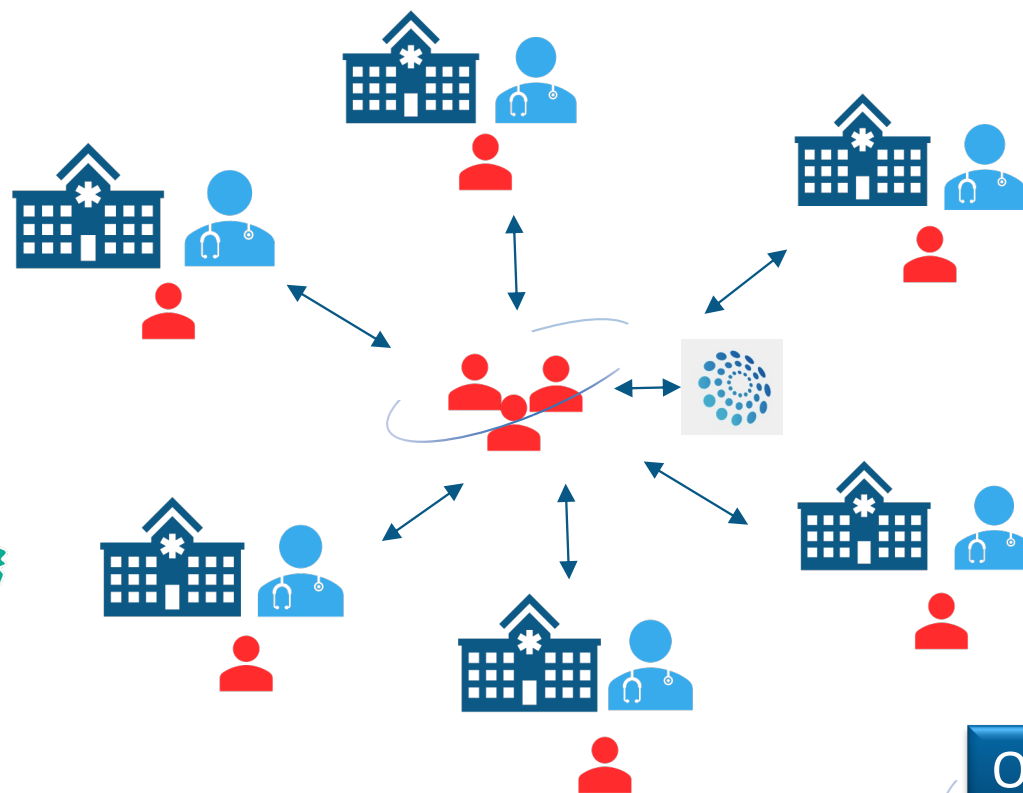
Indicators for the French National Plan

- a. Percentage of patients addressed to the expert centres by GPs over the years
- b. Percentage of diagnosis performed for new patients by year
- c. Impact of COVID Health Crisis on RD activities

<http://www.bndmr.fr/publications/presentations-communications/>

What can be done?

The Orphanet Data for RD proposed contribution



OD4RD

Ready to work with you!

- Orphanet is a well-established network on information and data: teams will be empowered as National Nomenclature Hubs
- ORPHAcodes are multi-lingual, interoperable, computable, and free
- Guidelines, files, tools, helpdesk developed in RD-CODE project
- Train-the-trainers programme developed in EJP RD to be scaled up
- Methodologies in place for collaboration with ERNs
- Lessons-learned from RD-CODE and EJP RD:
 - working hand-in-hand is the more accurate and straightforward way to go
- **But it needs willingness and means!**



THANK YOU!

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